RAI1 gene

retinoic acid induced 1

Normal Function

The *RAI1* gene provides instructions for making a protein that is active in nerve cells in the brain. Although the function of this protein is unknown, it is thought to be involved in nervous system development. Within cells, the RAI1 protein may be part of a protein complex (a group of proteins that work together) that helps control the activity of certain genes.

Health Conditions Related to Genetic Changes

Smith-Magenis syndrome

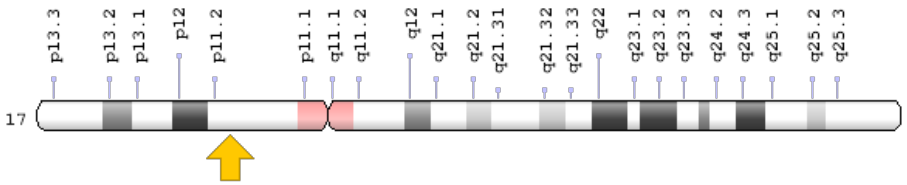
Researchers believe that a change in the function of the *RAI1* gene is responsible for most of the physical, mental, and behavioral features of Smith-Magenis syndrome. In most cases, one copy of the *RAI1* gene in each cell is lost when a region of chromosome 17 that contains the gene is deleted. It is unclear how the loss of this gene leads to the characteristic signs and symptoms of this condition.

A small percentage of cases are caused by a mutation in the *RAI1* gene instead of a chromosomal deletion. Although these individuals have many of the major features of Smith-Magenis syndrome (including intellectual disability, behavioral problems, and sleep disturbances), they are less likely than people with a chromosomal deletion to have short stature, hearing loss, and heart or kidney abnormalities. At least 12 *RAI1* mutations have been identified in people with Smith-Magenis syndrome. These mutations lead to the production of a nonfunctional version of the RAI1 protein or reduce the amount of this protein that is produced in cells. Scientists are working to determine how a missing or abnormal RAI1 protein results in the varied features of this disorder.

Chromosomal Location

Cytogenetic Location: 17p11.2, which is the short (p) arm of chromosome 17 at position 11.2

Molecular Location: base pairs 17,681,376 to 17,811,453 on chromosome 17 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- KIAA1820
- RAI1_HUMAN
- SMCR
- SMS

Additional Information & Resources

GeneReviews

- Smith-Magenis Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1310

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28RAI1%5BTIAB%5D%29+OR+%28retinoic+acid+induced+1%5BTIAB%5D%29%29+OR+%28%28SMCR%5BTIAB%5D%29+OR+%28KIAA1820%5BTIAB%5D%29+OR+%28MGC12824%5BTIAB%5D%29+OR+%28DKFZP434A139%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- RETINOIC ACID-INDUCED GENE 1
  http://omim.org/entry/607642

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=RAI1%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=9834
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/10743
- UniProt
  http://www.uniprot.org/uniprot/Q7Z5J4

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